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Genetics BIO2450 E158 Tuesday
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Lab Report #1: Cytogentics

Introduction: 'Case Study C' looks at 41 year old female patient in her seventeenth week of her pregnancy. Due to her age, the patient's child is at a higher risk for birth defects. The patient agreed to undergo a procedure called amniocentesis in which a sample of amniotic fluid is obtained and analyzed. The cells collected contain fetal skin cells that have been shed which are then cultured and arrested in metaphase of mitosis. The cells are then lysed and giemsa stained so that they can be observed under a light microscope and photographed.

Methods

Materials:

1. Chromoscan Board
2. Diagnostic Table

Method:

1. To create a karyotype correctly match the chromosome decals to the chromosomes on the Chromoscan Board to create homologous pairs.
2. When all chromosome decals have been placed analyze the karyotype for any abnormalities. Note inversions, translocations, deletions, insertions, or duplications.
3. Using the diagnostic table, diagnose the patient based on the obtained findings.
4.

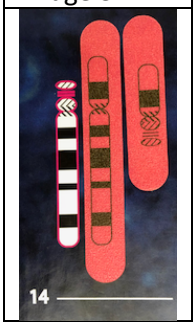
Results:

Image 1 shows the karyotype of 'Case Study C' . While analyzing out the karyotype of the amniotic fluid collected from 'Case Study C', it was observed that the fetal cells contained the normal number of chromosomes, 46 total. The unborn child is male which can be detected by looking at the sex chromosomes where there is are XY chromosomes. They SRY allele on the Y chromosome codes for males. Upon further analysis, an abnormality in the genotype was located at chromosome 14. At chromosome 14 there is a Robertsonian translocation of the p-arm from chromosome 21. The phenotype of this mutation results in a diagnosis of Down Syndrome. Image 2 shows the homologous pair of chromosomes 14 and image 3 shows the translocated p-arm of chromosome 21 next to the pair for reference.

Image 1

Image 2

Image 3



Discussion: Advanced maternal age has been pointed to as a factor for many complications for both fetus/baby and mother during pregnancy as well as genetic abnormalities expressed in phenotypes in children born to women over the age of 35. As a result, many pregnant women over the age of 35 chose to undergo testing to see if their fetus is healthy. Robertsonian translocations has a prevalence rate of 0.1% in the general population. According to a 2010 study, Robertsonian translocation are the most common structural chromosomal abnormality in the population. Among these translocations they occur mainly at 12,14,15,21,22 chromosomes. In a Robertsonian translocation two acrocentric chromosomes fuse at the centromere and lose their short arm. The diagnosis for this mutation is Down Syndrome. In 'Case Study C" we see at 14, 21 Robertsonian translocation. As a parent, or parent-to-be, genetic abnormalities can lead to a challenging decision. About 15%-20% of pregnancies end in abortion most often due to chromosomal abnormalities.

References

1. Blair, Christopher *BIO2450L Genetics Laboratory Manual;* New York City College of Technology; Brooklyn, NY
2. Ananthapur Venkateshwari, Avvari Srilekha, Tella Sunitha, Nallari Pratibha, and Akka Jyothy; *A Robertsonian Translocation rob (14;15) (q10:q10) in a Patient with Recurrent Abortions: A Case Report*, https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3719299/; Received 2010 Jun 19; Accepted 2010 Aug 24.